GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)

GepheID
GP00002236

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)

Trait State in Taxon A
Valais Blacknecked goat ; black forequarters and white hindquarters

Trait State in Taxon B
brown instead of black forequarters in Coppernecked goat

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name
goat

Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name
goat

Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Tyrp1

Synonyms
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1

String
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0004497 : monooxygenase activity

UniProtKB Mus musculus
P07147 (http://www.uniprot.org/uniprot/P07147)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0004497)

GO - Biological Process

GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0048023 : positive regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048023)
GO:0006583 : melanin biosynthetic process from tyrosine
(https://www.ebi.ac.uk/QuickGO/term/GO:0006583)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043438 : acetoacetic acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043438)
GO:0006582 : melanin metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006582)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030669)
GO:0010008 : endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0010008)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1487G>A p.Gly496Asp

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Asp	496

Main Reference

The brown coat colour of Coppernecked goats is associated with a non-synonymous variant at the TYRP1 locus on chromosome 8. (2015) (https://pubmed.ncbi.nlm.nih.gov/25392961)

Authors

Becker D; Otto M; Ammann P; Keller I; DrÃ¶gemÃ¼ller C; Leeb T

Abstract

The recent development of a goat SNP genotyping microarray enables genome-wide association studies in this important livestock species. We investigated the genetic basis of the black and brown coat colour in Valais Blacknecked and Coppernecked goats. A genome-wide association analysis using goat SNP50 BeadChip genotypes of 22 cases and 23 controls allowed us to map the locus for the brown coat colour to goat chromosome 8. The TYRP1 gene is located within the associated chromosomal region, and TYRP1 variants cause similar coat colour phenotypes in different species. We thus considered TYRP1 as a strong positional and functional candidate. We resequenced the caprine TYRP1 gene by Sanger and Illumina sequencing and identified two non-synonymous variants, p.Ile478Thr and p.Gly496Asp, that might have a functional impact on the TYRP1 protein. However, based on the obtained pedigree and genotype data, the brown coat colour in these goats is not due to a single recessive loss-of-function allele. Surprisingly, the genotype distribution and the pedigree data suggest that the (496) Asp allele might possibly act in a dominant manner. The (496) Asp allele was present in 77 of 81 investigated Coppernecked goats and did not occur in black goats. This strongly suggests heterogeneity underlying the brown coat colour in Coppernecked goats. Functional experiments or targeted matings will be required to verify the unexpected preliminary findings.

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Additional References

RELATED GEPHE

Related Genes

3 (Agouti (ASIP), EDNRA, MC1R) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9925^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001249/9925/